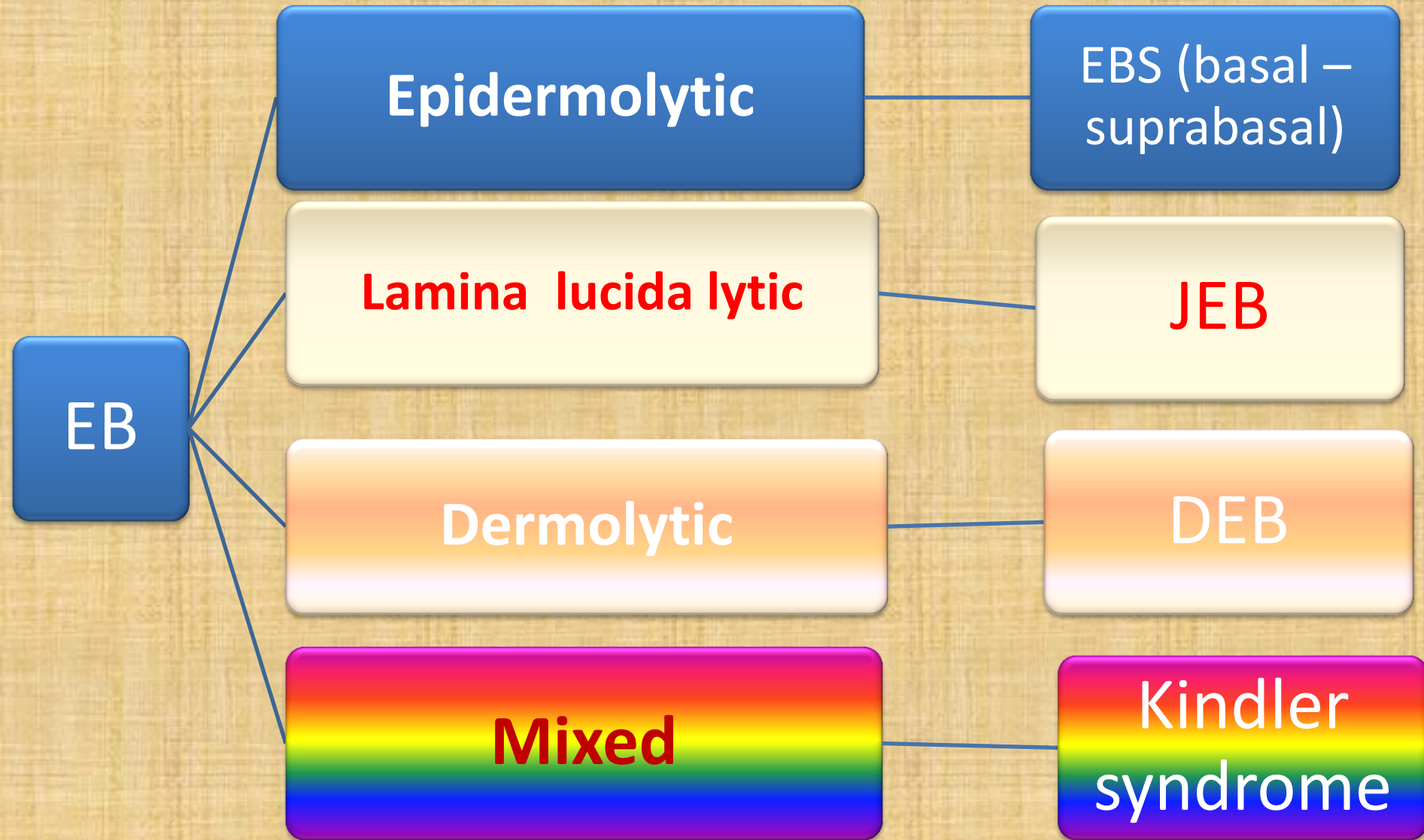


Classification of genetic epidermolysis bullosa

By

Prof. Dr. Essam Bakr

Revised classification system for epidermolysis bullosa (2008)



EPIDERMOLYTISIS BULLOSA SIMPLEX

• Supra basal

1. Plakophilin deficiency
2. Lethal acantholytic EBS
3. EBS superficialis

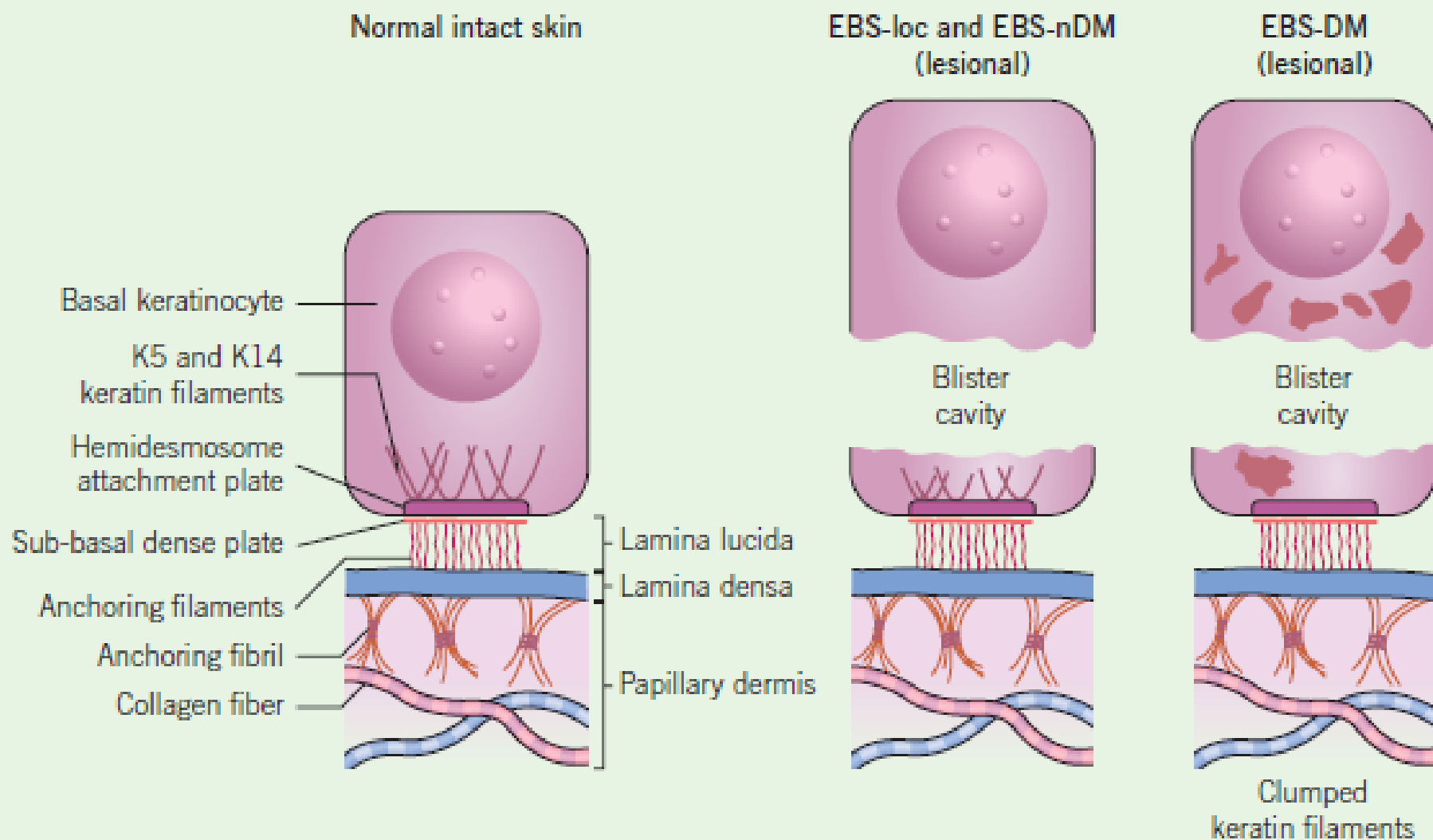
• Basal

1. Localised EBS
2. EBS, Dowling-Meara
3. EBS, Generalized other (Koebner)
4. EBS, AR
5. EBS, with Mottled hyperpig.
6. EBS, with Muscular dystrophy
7. EBS, Ogna
8. EBS, Migratory circinate
9. EBS, with Pyloric atresia

EB subtypes		Targeted gene product(s)
Plakophilin deficiency		plakophilin-1
Lethal acantholytic EBS		desmoplakin
EBS superficialis		unknown
Localized EBS		keratins 5 & 14
EBS, Dowling-Meara		keratins 5 & 14
EBS, generalized other		keratins 5 & 14
EBS, autosomal recessive		keratin 14
EBS with mottled hyperpigmentation		keratin 5
EBS with muscular dystrophy		plectin
EBS, Ogna		plectin
EBS, migratory circinate		keratin 5
EBS with pyloric atresia		$\alpha 6 \beta 4$ integrin, plectin

A

Ultrastructural site of blister formation in three major subtypes of EB simplex



Junctional epidermolysis bullosa

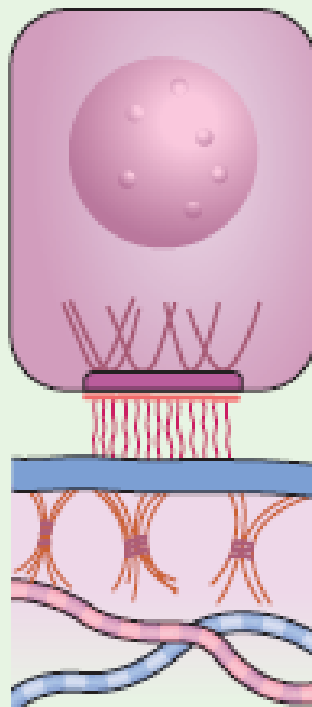
- All variants AR inheritance
- Blister formation at the level of Lamina lucida
- Types
 1. Herlitz JEB
 2. Non Herlitz JEB
 3. JEB, with pyloric atresia
 4. JEB, inversa
 5. JEB, late onset
 6. LOC Syndrome



B

Ultrastructural site of blister formation in two major subtypes of junctional EB

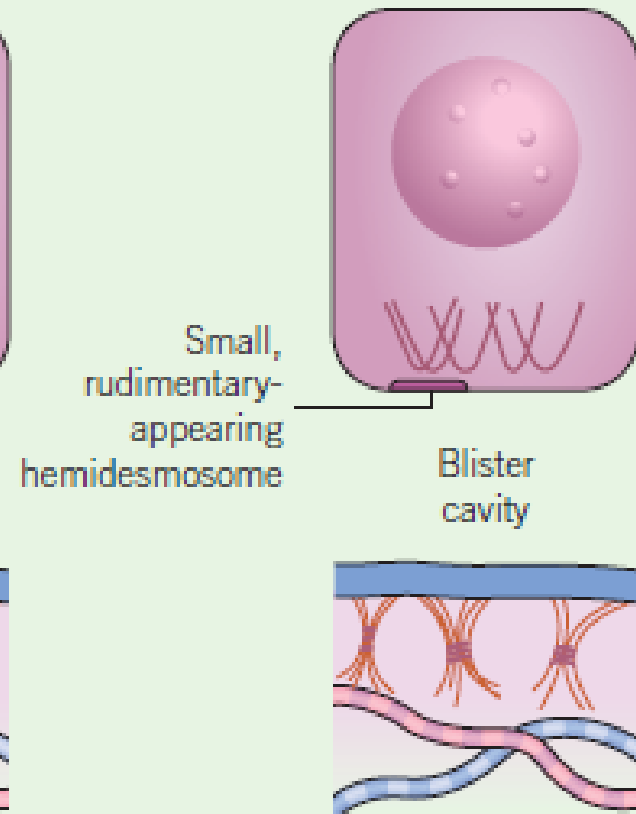
Normal intact skin



JEB-H (lesional)



JEB-nH (lesional)



continuous lamina densa in lower part.

Dystrophic Epidermolysis Bullosa

- Characterized by skin fragility, blistering, scarring, nail changes and milia formation.
- Unlike junctional EB, there are both autosomal recessive and autosomal dominant subtypes

Types

- Recessive DEB

- RDEB, severe generalized (*Hallopeau Siemens*)
- RDEB, generalized, other (*Non Hallopeau Siemens*)
- RDEB, inversa
- RDEB, pruriginosa
- RDEB, centripetalis
- RDEB, bullous dermolysis of the newborn

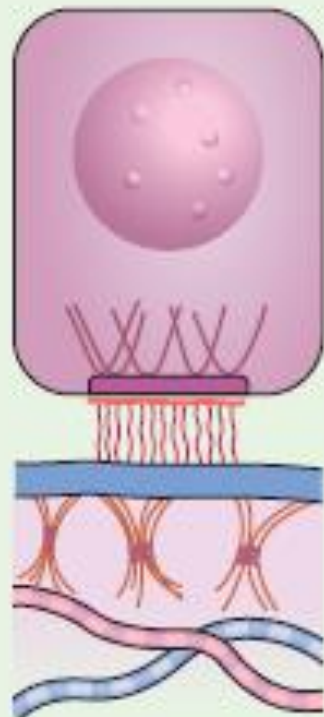
- Dominant DEB

- DDEB, generalized (Cockayne–Touraine) & (Pasini)
- DDEB, acral
- DDEB, pretibial
- DDEB, pruriginosa
- DDEB, nails only
- DDEB-bullous dermolysis of the newborn

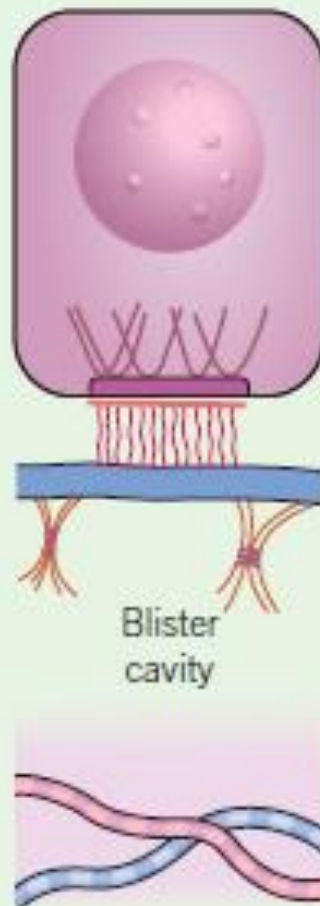
C

Ultrastructural site of blister formation in three major subtypes of dystrophic EB

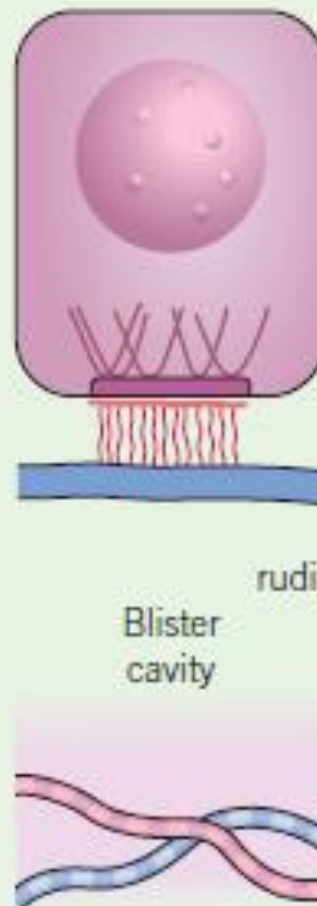
Normal intact skin



DDEB (lesional)



RDEB-sev/gen (lesional)



RDEB-o (lesional)

